



SIL1 gene

SIL1 nucleotide exchange factor

Normal Function

The *SIL1* gene provides instructions for producing a protein located in a cell structure called the endoplasmic reticulum. Among its many functions, the endoplasmic reticulum folds and modifies newly formed proteins so they have the correct 3-dimensional shape. The SIL1 protein works with BiP, a protein that helps fold newly produced proteins into the proper shape and refold damaged proteins. To start this process, BiP attaches (binds) to a molecule called adenosine triphosphate (ATP). When BiP folds a protein, the ATP is converted to a similar molecule called adenosine diphosphate (ADP). Then the SIL1 protein releases ADP from BiP so that it can bind to another molecule of ATP and start the protein folding process again. Because of its role in helping BiP exchange ADP for ATP, the SIL1 protein is called a nucleotide exchange factor.

Health Conditions Related to Genetic Changes

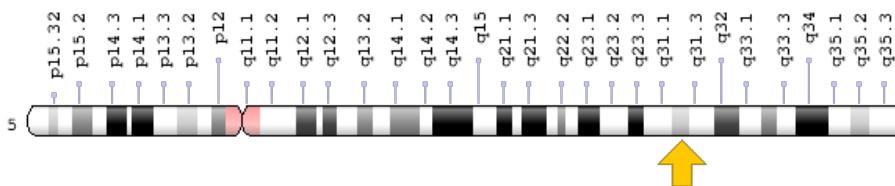
Marinesco-Sjögren syndrome

More than a dozen mutations in the *SIL1* gene have been found to cause Marinesco-Sjögren syndrome. Most of these mutations result in the production of a protein that has little or no activity. Defective SIL1 protein cannot remove ADP from BiP. BiP is then unable to bind to ATP and re-start the protein folding process. A disruption in protein folding impairs protein transport and causes proteins to accumulate in the endoplasmic reticulum. This accumulation likely damages and destroys cells in many different tissues, leading to poor coordination, muscle weakness, and the other features of Marinesco-Sjögren syndrome.

Chromosomal Location

Cytogenetic Location: 5q31.2, which is the long (q) arm of chromosome 5 at position 31.2

Molecular Location: base pairs 138,946,720 to 139,198,376 on chromosome 5 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- BAP
- BiP-associated protein
- MSS
- SIL1 homolog, endoplasmic reticulum chaperone (*S. cerevisiae*)
- SIL1 protein
- SIL1_HUMAN
- ULG5

Additional Information & Resources

Educational Resources

- Eurekah Bioscience Collection: The Discovery of Hsp70 Nucleotide Exchange Factors in Eukaryotes: Fishing Pays Off
<https://www.ncbi.nlm.nih.gov/books/NBK5987/#A61022>

GeneReviews

- Marinesco-Sjogren Syndrome
<https://www.ncbi.nlm.nih.gov/books/NBK1192>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SIL1%5BALL%5D%29+OR+%28BiP-associated+protein%5BALL%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+360+days%22%5Bdp%5D>

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- SIL1, S. CEREVISIAE, HOMOLOG OF
<http://omim.org/entry/608005>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SIL1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SIL1%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=24624
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/64374>
- UniProt
<http://www.uniprot.org/uniprot/Q9H173>

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